

Autoimmune Issues in the GI System

Our immune system can normally tell apart foreign entities and components of our own body. Sometimes, our immune system attacks parts of our own healthy body by mistake, resulting in **autoimmune disorders**. This info sheet will focus on these autoimmune GI diseases: **celiac disease**, **inflammatory bowel disease**, and **autoimmune enteritis**.

Celiac Disease

- The villi (finger-shaped projections) in the small intestines usually absorb nutrients from the food you eat, so that the rest of your body can use them.
- **Celiac disease occurs when the immune system attacks the gluten in the small intestine.** The villi gets damaged, flattened, and cannot absorb nutrients (see image).
- Celiac disease is genetic and can be passed onto children.
- Symptoms of celiac disease include (but are not limited to):
 - Frequent diarrhea
 - Tiredness
 - Constipation
 - Weight loss / failure to thrive
 - Pale, stinky stools
 - Anemia (not enough iron)
 - Bloating and gas
 - Nausea & vomiting
- The two main diagnostic tests are:
 - Blood test to search for antibodies against gluten
 - Genetic testing for HLA-DQ2 and HLA-DQ8, which are gene versions that make celiac disease more likely
 - **If you remove gluten from your diet before your diagnostic tests, you may have normal results even when you actually have celiac disease.**
- If you indeed have celiac disease, your doctor may check:
 - The severity of the damage in the small intestine. This is done by endoscopy – sending a tiny camera into your digestive tract using a long thin tube and taking a small biopsy sample of the inside of your intestine.
 - Whether you are lacking certain vitamins, minerals, and supplements. This is done by a blood test.
- The only way to prevent the immune system from attacking the small intestine is to **stop eating gluten for the rest of your life**. Patients who continue to eat gluten will have malnutrition, which can lead to long lasting problems for the brain, nerves, bones, and muscles.
- Stopping gluten will allow the intestines to heal, which happens in a few months to a few years in most cases. Supplementation of vitamins (e.g. B12 and D) and minerals (e.g. iron) may be needed initially. For the 5% of celiac disease patients whose intestines do not recover, special care is needed.

Autoimmune GI diseases in individuals with 22q11.2DS

- In children with 22q11.2DS, celiac disease, inflammatory bowel disease, and autoimmune enteritis are relatively rare.
- Among adults with 22q11.2DS, celiac disease and inflammatory bowel disease occur at frequencies similar to those of the general population.



Inflammation of the mucous layer of the small intestine may lead to the flattening of the villi. Source: <https://www.scientificanimations.com/wiki-images/> under Creative Commons "BY-SA (Attribution-ShareAlike 4.0 International)" license

Gluten

Gluten is a protein in wheat, barley, rye, and mixed oats. Products that contain gluten include (but are not limited to):

- Most bread, pastries, cereals, pasta, communion wafers, and some alcoholic drinks
- Many sauces and processed foods
- Some preservatives, vitamins, supplements, medications, makeup, toothpaste and mouthwash, playdough, and glue on envelope/stamps

Autoimmune Issues in the GI System (continued)

Inflammatory Bowel Disease (IBD)

- IBD is a group of disorders that involve long-term inflammation (pain and swelling) of the GI tract.
- Environmental triggers, immune responses, and genetic predisposition may be some of the factors that cause enteropathy (diseases in the intestines).
- There are two main types:
 - **Ulcerative colitis** causes swelling and ulcers (sores) in the colon and rectum.
 - **Crohn's disease** affects any part of the GI tract, from the mouth to the anus.
- Symptoms include (but are not limited to):
 - Abdominal pain
 - Diarrhea (+ blood)
 - Bleeding from rectum
 - Low appetite
 - Weight loss
 - Fatigue
- Some tests that may help diagnose IBD:
 - Blood tests
 - Stool sample analysis
 - Small bowel ultrasound or MRI studies
 - Endoscopy / Colonoscopy – sending a tiny camera into the GI tract to check the interior
- Treatment for IBD may include:
 - Using medications to suppress the immune reactions or deal with the swelling
 - Mediterranean diet, Crohn disease exclusion diet, or specific carbohydrate diet
 - Performing surgery to remove the affected part of the intestines (if medications do not help)
 - Supplementation of vitamins (e.g. B12, D) and minerals (e.g. iron) that are deficient as a result of the inflammation
- Unmanaged IBD can cause severe damage to the intestines and increase the risk of colon cancer. The inflammation can also affect other areas of the body.

Note: Inflammatory bowel disease IBD is not the same as [irritable bowel syndrome \(IBS\)](#).

Autoimmune Enteropathy/Enteritis

- Autoimmune enteropathy happens when the immune system damages the lining of the intestines. The patient has severe diarrhea very frequently.
- These symptoms can start happening soon after birth:
 - Having loose poop all the time, sometimes with blood in the poop
 - Poor weight gain
 - Not peeing often
 - Other infections
 - Skin rashes
 - Problem with blood sugar levels
 - Imbalances of electrolytes including (but not limited to) sodium, potassium, magnesium, chloride etc.
- Get medical help if these symptoms are present
 - Dehydration
 - Diarrhea becomes worse
 - Fever (temperature over 100.4°F or 38°C)
- Some tests that may help diagnose enteropathy:
 - Blood tests
 - Endoscopy / Colonoscopy – sending a tiny camera into the GI tract to check the interior
- Treatment:
 - Giving the patient fluids and nutrition intravenously (into the vein)
 - Special diet
 - Medications to suppress the immune system

References / Resources

Clinical practice recommendations and research reports

- [Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome](#) – 2023
- [Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome](#) – 2023
- [Gastrointestinal Features of 22q11.2 Deletion Syndrome Include Chronic Motility Problems From Childhood to Adulthood](#) – 2022

Websites of medical institutions

- [Celiac Disease](#) | [Inflammatory Bowel Disease](#) – Johns Hopkins Medicine
- [Celiac Disease](#) | [Inflammatory Bowel Disease](#) – Mayo Clinic
- [Celiac Disease](#) | [Inflammatory Bowel Disease](#) – Cleveland Clinic
- [Celiac Disease](#) | [Ulcerative Colitis](#) | [Crohn's Disease](#) – GI Kids by the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (NASPGHAN)
- [Autoimmune Enteropathy](#) – Children's Hospital of Philadelphia (CHOP)
- [Autoimmune Enteropathy](#) - Cincinnati Children's Hospital